FACT SHEET
Healthcare Provider

Multiple CoA Carboxylase Deficiency (MCD)

Description:
Multiple CoA Carboxylase Deficiency is an autosomal recessive metabolic disorder caused by a deficiency of the enzyme holocarboxylase synthase (HS). This deficiency leads to impaired activity of three enzymes that are dependent on biotin, part of the Vitamin B complex. Holocarboxylase synthetase (HS) attaches biotin to the four-carboxylase enzymes (pyruvate carboxylase, propionyl CoA carboxylase, beta-methylcrotonyl CoA carboxylase, and acetyl CoA carboxylase) in order to activate them. Deficiency in HS results in a functional decrease in the enzymatic activity of biotin-dependent carboxylases.

Incidence in General Population:
1:100,000 live births

Symptoms:
Symptoms include seizures, ketoacidosis, hypotonia, immune system impairment, a diffuse erythematous rash, alopecia, hearing loss and developmental retardation. The disorder occurs in both a neonatal and late-onset form and is treatable. Infants generally present with food refusal, vomiting, breathing problems, hypotonia, seizures, and lethargy. Severe metabolic/lactic acidosis, organic aciduria, mild hyperammonemia and variable hypoglycemia can lead to coma and death if not treated. Survivors can have neurological damage.

Diagnosis:
Newborn screening abnormality—Tandem mass spectrometry: increased C5OH.
A second dried blood spot filter paper card may be requested by the Newborn Screening Laboratory if the initial screening result is above the normal range. Infants with presumptive positive screening (critical) results require prompt follow up. If this occurred, the clinician would be contacted by the Metabolic Treatment Center. When notified of these results, the clinician should immediately check on the clinical status of the baby and facilitate referral to the Metabolic Treatment Center. The Metabolic Treatment Center will provide consultation and assistance with diagnostic testing.

Treatment:
Treatment is oral biotin supplementation, which should begin immediately upon diagnosis. Majority of cases respond readily to biotin supplementation. Biotin increases the functional activation of the carboxylase enzymes.

Immunization:
Immunizations must be kept current. Influenza vaccinations are also recommended.

Growth and development:
Children with holocarboxylase synthetase deficiency treated with biotin can have normal growth and development. Biotin supplementation should be maintained through the lifetime of the affected individual. However, some partly respond to only therapy and one has been reported to be unresponsive to biotin therapy.

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